

# HGEN 6500: Human Genetics

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**Credit Hours: 3.0**

## Contact Information

Name	Position	Phone	Email
Faculty TBD			UUGPGC@utah.edu

## Course Information

### Brief Description of Course

This course covers topics related to human genetics and the mechanisms of gene and cellular regulation, DNA structure, genes and mutations, biology of genetic disorders, patterns of inheritance, basic cytogenetics, population genetics, and risk analysis. This course consists of lectures by the instructor(s) and by guest lecturers. Prerequisite: University of Utah Graduate Program in Genetic Counseling (UUGPGC) students or permission of instructor; undergraduate course in genetics.

### Course Objectives

At the conclusion of this course students will be able to:

1. Explain and apply important concepts in biology to medicine.
2. Describe the causes and clinical consequences of mutation, particularly in terms of variation in the clinical expression of genetic disorders.
3. Determine the risk for an individual to inherit a genetic condition and/or develop disease based on the presence of a mutation or a positive family history; this includes applying Bayesian analysis when applicable.
4. Recognize concepts of population genetics, including Hardy-Weinberg equilibrium, founder effects, heterozygous advantage, and population
5. Describe and illustrate techniques of molecular analysis and their application to genetic disease diagnoses. This includes nomenclature for DNA sequence variants, analyses of pathogenicity of sequence variants, and principles of genetic counseling related to client communication of molecular results.
6. Recognize and apply the basic concepts of cytogenetics including nomenclature, features of meiosis and mitosis, numerical abnormalities of chromosomes and their clinical consequences, and techniques of cytogenetic analysis and their application to genetic disease.
7. Explain the concepts of Mendelian traits versus complex non-Mendelian traits within the context of human populations and discuss multifactorial genetic susceptibility for birth defects and chronic.
8. Critically interpret genetic test results in the context of clinical phenotype and family
9. Outline applications of genomics-based precision medicine and describe therapeutic interventions for genetic condition.

### The ACGC Practice-Based Competencies Addressed in this course:

- Genetics and Genomics Expertise: 1; 1.a; 1.b; 1.c
- Risk Assessment: 2; 2.a; 2.b; 2.c
- Communication: 4
- Healthcare Systems: 6.a
- Professional Identity: 7; 7.a; 7.b; 7.c

## Course Format & Schedule

### Timeline

One semester

### Educational and Instructional Modalities

This course is held in person for two 90-minute sessions a week. There will be some asynchrony work and occasional recorded lectures within class discussion.

**Role of the Student in this Course**

You will be expected to engage in a variety of learning activities including reading, viewing videos, sharing with peers, writing and creating educational presentations.

**Required Textbooks/Readings**

- Laptop or tablet with a webcam & microphone (or headphones)
- Required Textbooks: Jorde L B, Carey JC, and Bamshad MJ. (2020) Medical Genetics (6th edition).
- Optional Textbooks: Reed and Donai (2015) Clinical Genetics (3rd edition)
- Assigned Readings, Optional Readings, Resources:
  - Readings will be found in Canvas under each individual class and may be printed or downloaded on your home computer. Make sure you read the headers and footers, as they contain textbook chapter information and websites that are required reading.

**Assessment & Grading**

**Evaluation Methods**

1. Exams: two examinations will be given (midterm and final examination) each worth 20% of the final grade. All students must take both exams. Exams cover lecture and homework content as well as readings from the textbook and assigned articles. The final examination is comprehensive and covers all semester material. A student will be given an early or late exam only in the event of an extreme emergency.
2. Homework and Quizzes: homework assignments and quizzes will account for 30% of the final grade. Please make sure that all your questions are answered during lectures to make sure you can complete the homework problems. On the due date, homework should be submitted electronically or in person by the specified time. These assignments will cover lecture and reading materials. \*One point, per day will be deducted for any assignments turned in late. Homework will be returned by either the Midterm or Final exam. Quizzes may be announced or unannounced.
3. Gene Function Assignment: Each student will be assigned a genetic condition at the beginning of the semester and will present that condition at the end of the semester based on personal research and knowledge gained during this course. This assignment will count for 20% of the overall grade.
4. Participation: Up to 10% of the final grade will be given for active participation. This means attending lectures, being prepared for class, and initiating and participating in class discussions. 3% of the total participation percentage will be awarded for the mid-year evaluation. This is an "all or none" credit, all students must submit the evaluation in order for everyone to receive the credit.

**Grading System**

Students will receive a final letter grade. To pass this course you must achieve an overall score of at least 80% (a B- equivalent) as required by the Department of Human Genetics.

Grading Scale

A	95-100
A-	90-94
B+	86-89
B	82-85
B-	79 – 81
C+	76-78
C	73-75
C-	70-72
D+	67-69
D	64-66
D-	60-63
F	Below 60



## Standard Policies

### **The Americans with Disabilities Act.**

The University of Utah seeks to provide equal access to its programs, services, and activities for people with disabilities. If you will need accommodations in this class, reasonable prior notice needs to be given to the Center for Disability Services, 162 Olpin Union Building, 801-581-5020. CDS will work with you and the instructor to make arrangements for accommodation. All written information in this course can be made available in an alternative format with prior notification to the Center for Disability Services.

### **Addressing Sexual Misconduct.**

Title IX makes it clear that violence and harassment based on sex and gender (which includes sexual orientation and gender identity/expression) is a civil rights offense subject to the same kinds of accountability and the same kinds of support applied to offenses against other protected categories such as race, national origin, color, religion, age, status as a person with a disability, veteran's status or genetic information. If you or someone you know has been harassed or assaulted, you are encouraged to report it to the Title IX Coordinator in the Office of Equal Opportunity and Affirmative Action, 135 Park Building, 801-581-8365, or the Office of the Dean of Students, 270 Union Building, 801-581-7066. For support and confidential consultation, contact the Center for Student Wellness, 426 SSB, 801-581-7776. To report to the police, contact the Department of Public Safety, 801-585-2677 (COPS).

### **Campus Safety.**

The University of Utah values the safety of all campus community members. To report suspicious activity, call campus police at 801-585-COPS (801-585-2677). You will receive important emergency alerts and safety messages regarding campus safety via text message. For more information regarding safety and to view available training resources, including helpful videos, visit [safeu.utah.edu](http://safeu.utah.edu).

### **Alternate Name and/or Personal Pronoun**

Class rosters are provided to the instructor with the student's legal name as well as 'Preferred' first name (if previously entered by you in the Student Profile section of your CIS account). While CIS refers to this as merely a preference, we will honor you by referring to you with the name and pronoun that feels best for you in class, on papers, exams, group projects, etc. Please advise us of any name or pronoun changes (and please update CIS) so we can help create a learning environment in which you, your name, and your pronoun will be respected.

### **Current COVID-19 Campus Guidelines**

COVID-19 Guidelines change rapidly. Please access the most current information COVID-19 Central @ The U. [coronavirus.utah.edu](http://coronavirus.utah.edu)